

46, XY, t (4q; 7q⁺) Translocation in Laurence-Moon-Bardet-Biedl Syndrome: A Case Report

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KEYWORDS Laurence-Moon-Bardet-Biedl Syndrome. Mental Retardation. Polydactyly. Obesity. Translocation

ABSTRACT A sixteen year old boy was brought to the hospital with complaints of poor vision. A few cardinal features of Laurence Moon Bardet Biedl syndrome were observed viz., central obesity, hypogonadism, retinitis pigmentosa, mental retardation, delay of the speech and polydactyly. Emphasis was placed on finding out the chromosomal aberrations that might serve as a diagnostic marker for the disease at a state when some of the characteristic features of the syndrome may be lacking. The significance of diagnosis and its importance in genetic counseling are discussed pertaining to the recent literatures.

INTRODUCTION

Laurence-Moon-Bardet-Biedl Syndrome (MIM 209900), first described in 1920, is an autosomal recessive condition with a wide spectrum of clinical features. LMBBS is a rare, genetically heterogeneous, autosomal recessive disorder characterized by early onset of retinitis pigmentosa, post axial polydactyly, central obesity, mental retardation, hypogonadism and renal anomalies (Schachat and Maumenee 1982).

The cardinal manifestations are retinal pigmentary dystrophy (previously termed retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, and hypogonadism. Minor features include hepatic fibrosis, diabetes mellitus, reproductive abnormalities, endocrinological disturbances, short stature, hearing loss, developmental delay, and speech deficit.

Other features that vary in frequency include diabetes mellitus, hypertension and congenital heart disease (Nishimura et al. 2001). Main aim of the present study was to find out the chromosomal aberrations if any that might help in diagnosis of the syndrome.

CASE REPORT

A sixteen year old boy was brought to the outpatient ward of Coimbatore Medical College Hospital with complaint of excessive weight gain noticed since early childhood and poor vision (Fig. 1). He was born out of non-consanguineous marriage of who were asymptomatic of the condition. He was born at term, with no antenatal or perinatal complications. The patient had a younger brother who was normal but the patient's sister had the same complications as of the patient. The patient had progressive deterioration in vision for the last five to six years, starting with difficulty in seeing at night. Systemic evaluation revealed truncal obesity (Fig. 1), polydactyly (post axial, present only in hands and not in legs) (Fig. 2a, b), truncal obesity, hypogonadism, micropenis (Fig. 3) and speech disorder. Laboratory investigations were normal. Serum tests were unrewarding.

Hence a diagnosis of Laurence-Moon-Bardet-Biedl syndrome was made based on physical observations (Table 1). It was decided to go for genetic analysis in the form of karyotyping to determine abnormal chromosomal aberrations if any.

To disclose the association of genetics with this disorder, the chromosomes of the patient were analyzed karyotyping on peripheral blood lymphocyte culture. Interestingly, a translocation of deleted long arm portion of 4th chromosome to the long arm of chromosome 7 (46, XY, t (4q-; 7q⁺) was observed (Fig. 4).

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Fig. 1. Central obesity of the patient

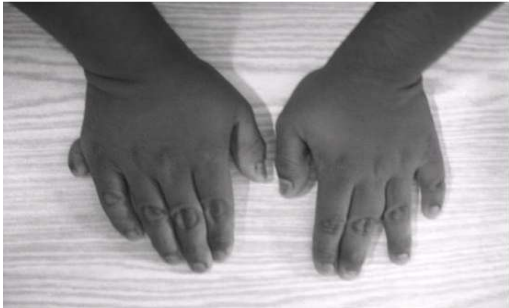


Fig. 2a. Polydactyl of right hand, extra finger in the left hand operated out during his childhood days



Fig. 2b. No polydactyly in legs



Fig. 3. Micropenis

Table 1: Characteristic features observed in the patient

Features	
<i>Primary Features</i>	
Red cone dystrophy	+
Post axial Polydactyl	+
Truncal obesity	+
Hypogonadism	+
Renal anomalies	-
<i>Secondary Features</i>	
Speech disorder/delay	+
Developmental delay	+
Behaviour	+
Ataxia/imbalance	-
Diabetes mellitus	-
Congenital heart defects	-
Liver disease	-
Hearing loss	-
Facial features	+
Situs inversus	-
Hirschprung disease	-
Polyuria/polydipsia	-
Dental crowding	+
Anosmia	-
Vision on left and Right eyes	Affected during night time.

DISCUSSION

The patient of our study is a classical case of Laurence-Moon-Bardet-Biedl syndrome. The case presented with several characteristic features of Laurence-Moon-Bardet-Biedl syndrome including the five cardinal features namely central obesity, hypogonadism, retinitis pigmentosa, mental retardation, delay of the speech and systemic abnormalities like polydactyly. Karyotypic analysis revealed a translocation of deleted long arm portion of 4th chromosome to the long arm of chromosome 7 (46, XY, t (4q- ; 7q⁺).

Badano et al. (2003) represented a deletion of 4q27 and Nishimura et al. (2005) demonstrated



Fig. 4. Metaphase plate 46, XY t (4q-; 7q+)

the deletion of a region 7p14 chromosome in Bardet Biedl syndrome. Bardet-Biedl syndrome (BBS) is also a genetically heterogeneous disorder, the primary features of which include obesity, retinal dystrophy, polydactyly, hypogonitalism, learning difficulties, and renal malformations (Badano et al. 2003).

Hence the present study results reveal that a chromosomal translocation between chromosomes 4 and 7 may aid in diagnosis for LMBBS in patients lacking the characteristic features of LMBBS. But a larger number of cases need to be studied in order to confirm this finding. Further, the genes residing on the long of chromosome 4 and 7 should be probed further for association with this genetic disorder.

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